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(71) Applicant (for all designated States except US): **UNIVERSITY OF CHICAGO** [US/US]; 5555 South Woodlawn Avenue, Suite 300, Chicago, IL 60637 (US).

(72) Inventors; and

(75) Inventors/Applicants (for US only): **RATAIN, Mark, J.** [US/US]; 1040 West Oakdale Avenue, Chicago, IL 60657 (US). **INNOCENTI, Federico** [—/US]. **KROETZ, Deanna, L.** [—/US]. **UNDEVIA, Samir.** **NGUYEN, Tan, D.** [US/—]; 511 Pennsylvania Avenue, San Francisco, CA 94107 (US). **LIU, Wanqing** [CN/—].

(74) Agent: **SHISHIMA, Gina, N.**; Fulbright & Jaworski, LLP, 600 Congress Avenue, Suite 2400, Austin, TX 78701 (US).

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(54) Title: METHODS AND COMPOSITIONS RELATING TO THE PHARMACOGENETICS OF ABCC2, UGT1A1, AND/OR SLC01B1 GENE VARIANTS

(57) Abstract: The present invention is directed to methods and compositions for determining the presence or absence of polymorphisms within an ABCC2, *UGT1A1*, and/or *SLC01B1* gene and correlating these polymorphisms with activity levels of their gene products and making evaluations regarding the effect on their substrates, particularly those substrates that are drugs. In addition, there are methods and compositions of evaluating the risk of an individual for developing toxicity or adverse event(s) to an ABCC2, UGT1A1, and/or SLC01B1 substrate. In some embodiments, the invention concerns methods and compositions for determining the presence or absence of ABCC2 3972C>T variant and predicting or anticipating the level of activity of ABCC2 and determining dosages of an ABCC2 drug substrate, such as irinotecan, in a patient. Such methods and compositions can be used to evaluate whether irinotecan-based therapy, or therapy involving other ABCC2 substrates, may pose toxicity problems if given to a particular patient or predicting their efficacy. Alterations in suggested therapy may ensue based on genotyping results.



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